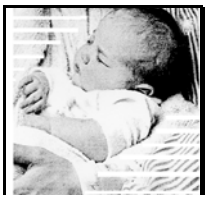

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

Medium Chain Acyl-CoA Dehydrogenase Deficiency is an autosomal recessive fatty acid oxidation disorder (FOD) in which an enzyme defect in the fatty acid metabolic pathway inhibits the body's ability to utilize stored fat. Clinical symptoms include vomiting and lethargy following a period of fasting, often at times of intercurrent viral infection (gastrointestinal or upper respiratory). Hypoglycemia with low urinary ketone production (hypoketotic), hyperammomemia and elevated liver function tests may occur and can lead to encephalopathy, hepatic failure, coma or death.

Incidence (MI):	Unknown Estimated between 1:10,000 and 1:15,000
Laboratory Screening Test:	Acylcarnitine profiling by Tandem Mass Spectrometry
Timing of Test:	Optimum time for detection is between 24-36 hours of age. Disorder could be missed if specimen collection is delayed.
Feeding Effect:	None
Transfusion Effect:	None
Confirmation:	All presumptive positive tests are referred to the Pediatric Neurology Metabolic Clinic (PNMC) for confirmation (734) 763-4697. **Do not send diagnostic labs before contacting the PNMC.**
Treatment:	Effective treatment for MCAD deficiency includes a low-fat diet and avoidance of fasting. The prescription drug carnitine is used. Acute episodes are managed by the administration of intravenous glucose

For further information, contact the Newborn Screening Program
Telephone (517) 335-9205 Fax (517) 335-9419
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